

**Table 5. Summary of Sequence Variations in *TGIF***

	<b>Sequence Change</b>	<b>Expected Effect</b>	<b>Type of Mutation</b>	<b>Reference</b>
<b>Mutations</b>	83C→G	S28C	Missense	<a href="#">Gripp et al 2000</a>
	188C→G	P63R	Missense	<a href="#">Gripp et al 2000</a>
	451A→G	T151A	Missense	<a href="#">Gripp et al 2000</a>
	485C→T	S162F	Missense	<a href="#">Gripp et al 2000</a>
	<b>Sequence Change</b>	<b>Expected Effect</b>	<b>Frequency</b>	<b>Reference</b>
<b>Polymorphisms</b>	371C→T	S20S	1/357	Present study
	420A→G	P140P	25/357	<a href="#">Nanni et al 2000</a>
	487C→T	P163S	22/357	<a href="#">Nanni et al 2000</a>
	488C→T	P163L	27/357	<a href="#">Nanni et al 2000</a>
	887C→T	V192V	4/357	Present study
	968T→G	T219T	6/357	Present study

## References

Gripp KW, Wotton D, Edwards MC, Roessler E, Ades L, Meinecke P, Richieri-Costa A, Zackai EH, Massague J, Muenke M, Elledge SJ (2000) Mutations in *TGIF* cause holoprosencephaly and link NODAL signalling to human neural axis determination. *Nat Genet* 25:205-8 [[Medline](#)]

Nanni L, Croen LA, Lammer EJ, Muenke M (2000) Holoprosencephaly: molecular study of a California population. *Am J Med Genet* 90:315-9 [[Medline](#)]